

## AMENDMENTS TO THE SPECIFICATION

Please insert the following Tables 1, 2, 3, 4, 5 and 6 after paragraph [0112] and before the Claims.

**Table 1**

**Number and Frequency (%) of *ACTN3* Genotypes and Frequency (%) of *ACTN3* Alleles in Controls and Elite Sprint/Power and Endurance Athletes**

<b>GROUP (n)</b>	<b>NO. (%) WITH GENOTYPE</b>			<b>ALLELE FREQUENCY (%)</b>	
	<b>RR</b>	<b>RX</b>	<b>XX</b>	<b>R</b>	<b>X</b>
<b>Males:</b>					
Controls (134)	40 (30)	73 (54)	21 (16)	57	43
Sprint (72)	38 (53)	28 (39)	6 (8)	72	28
Endurance (122)	34 (28)	63 (52)	25 (20)	54	46
<b>Females:</b>					
Controls (292)	88 (30)	147 (50)	57 (20)	55	45
Sprint (35)	15 (43)	20 (57)	0 (0)	71	29
Endurance (72)	26 (36)	25 (35)	21 (29)	53	47
<b>Total:</b>					
Controls (436)	130 (30)	226 (52)	80 (18)	56	44
Sprint (107)	53 (50)	48 (45)	6 (6)	72	28
Endurance (194)	60 (31)	88 (45)	46 (24)	54	46

**TABLE 2: Genotyping of R577X in ACTN3 in Caucasians Elite Athletes.**

Strength	Sport	ID	Sport Institute	Total Number	577RR (%)	577RX (%)	577XX (%)
Endurance	Rower	RT492	AIS	64	22	28	14
		- RT556			(34.4%)	(43.8%)	(21.8%)
Endurance	Triathlete	RT977	AIS	13	3	8	2
		- RT989			(23.1%)	(61.5%)	(15.4%)
Endurance	Cyclist	RT990	AIS	9	4	2	3
		- RT998			(44.4%)	(22.2%)	(33.3%)
Endurance	Track Cyclist	KN246	AIS	22	7	7	8
		- KN275			(31.8%)	(31.8%)	(36.4%)
Endurance	Marathon	KN310	AIS	1	0	0	1
Endurance	All above		AIS	108	36 (33.3%)	45 (41.7%)	27 (25.0%)
Sprint	Swimmer	RT901	AIS	45	17	25	3
		- RT1018			(37.8%)	(55.6%)	(6.6%)
Sprint	Track Cyclist	KN246	AIS	8	4	3	1
		- KN275			(50.0%)	(37.5%)	(12.5%)
Sprint	Athletics	KN276	AIS	30	16	13	1
		- KN309			(53.3%)	(43.3%)	(3.3%)
Sprint	All above		AIS	83	37 (44.6%)	41 (49.4%)	5 (6.0%)
Africa Zulu				88	69 (78.4%)	18 (20.5%)	1 (1.1%)
Australian Caucasian Control				152	46 (30.0%)	78 (52.0%)	28 (18%)

**Table 3** SNPs identified in the ACTN3 gene to date  
NCBI SNP CLUSTER ID

rs2229456  
rs2229455  
rs2229454  
rs2228325  
rs1126675  
rs7949754  
rs7924602  
rs5792393  
rs4990284  
rs4990283  
rs4013815  
rs3937320  
rs3837428  
rs3814736  
rs3814735  
rs3782080  
rs2511217  
rs2511216  
rs2509559  
rs2509558  
rs2305537  
rs2305534  
rs2290463  
rs2275998  
rs2096583  
rs2000939  
rs1815739  
rs1791690  
rs1671064

rs679228  
rs678397  
rs677488  
rs647476  
rs647029  
rs618838  
rs607736  
rs597626  
rs544021  
rs540874  
rs538330  
rs531490  
rs509556  
rs490998  
rs13897  
rs4576  
rs1189338  
rs1201433  
rs640213  
rs3737525  
rs3178740  
rs3180065  
rs3180064  
rs3180063  
rs3867132  
rs608504  
rs610293  
rs3825065

**TABLE 4.** Symbols, full names, and cytogenic location of nuclear and mitochondrial genes of the 2002 Human Gene Map for Performance and Health-Related Fitness Phenotypes.

**Gene or Locus Name Location**

**A B**

**ACADVL** Acyl coenzyme A dehydrogenase, very long chain 17p13-p11

**ACE** Angiotensin I converting enzyme 17q23

**ADRA2A** Alpha-2A-adrenergic receptor 10q24-q26

**ADRB1** Adrenergic, beta-1-, receptor 10q24-q26

**ADRB2** Beta-2-adrenergic receptor 5q31-q32

**ADRB3** Beta-3-adrenergic receptor 8p12-p11.2

**AGT** Angiotensinogen 1q42-q43

**ANG** Angiogenin, ribonuclease, RNase A family, 5 14q11.1-q11.2

**APOE** Apolipoprotein E 19q13.2

**ATP1A2** ATPase, Na<sub>2</sub>/K<sub>2</sub> transporting, alpha-2 polypeptide 1q21-q23

**ATP1B1** ATPase, Na<sub>2</sub>/K<sub>2</sub> transporting, beta 1 polypeptide 1q22-q25

**BDKRB2** Bradykinin receptor B2 14q32.1-q32.2

**C D E F G**

**CASQ2** Calsequestrin 2 (cardiac muscle) 1p13.3-p11

**CFTR** Cystic fibrosis transmembrane conductance regulator, ATP-binding cassette (sub-family C, member 7) 7q31.2

**CKM** Creatine kinase, muscle 19q13.2-q13.3

**CNTF** Ciliary neurotrophic factor 11q12.2

**CPT2** Carnitine palmitoyltransferase 2 1p32

**COL1A1** Collagen, type I, alpha 1 17q21.3-q22.1

**EDN1** Endothelin 1 6p24.1

**ENO3** Enolase 3, (beta, muscle) 17pter-p11

**FABP2** Fatty acid binding protein 2 4q28-q31

**FGA** Fibrinogen, A alpha polypeptide 4q28

**FGB** Fibrinogen, B beta polypeptide 4q28

**GDF8 (MSTN)** Growth differentiation factor 8 (myostatin) 2q32.2

**GNB3** Guanine nucleotide binding protein (G protein), beta polypeptide 3 12p13

**H I K L M**

**HLA-A** Major histocompatibility complex, class I, A 6p21.3

**HP** Haptoglobin 16q22.1

**IGF1** Insulin-like growth factor I 12q22-q23

**IGF2** Insulin-like growth factor 2 11p15.5

**IL-6** Interleukin-6

**KCNQ1** K<sub>2</sub> voltage-gated channel, KQT-like subfamily, member 1 11p15.5

**LDHA** Lactate dehydrogenase A 11p15.4

**LPL** Lipoprotein lipase 8p22

**MTCO1** Cytochrome c oxidase I mtDNA 5904-7445

**MTCO3** Cytochrome c oxidase III mtDNA 9207-9990

**MTCYB** Cytochrome b mtDNA 14747-15887

**MTND1** NADH dehydrogenase 1 mtDNA 3307-4262

**MTND4** NADH dehydrogenase 4 mtDNA 10760-12137

***MTND5*** NADH dehydrogenase 5 mtDNA 12337–14148  
***MTTE*** Transfer RNA, mitochondrial, glutamic acid mtDNA 14674–14742  
***MTTI*** Transfer RNA, mitochondrial, isoleucine mtDNA 4263–4331  
***MTTK*** Transfer RNA, mitochondrial, lysine mtDNA 8295–8364  
***MTTL1*** Transfer RNA, mitochondrial, leucine 1 (UUR) mtDNA 3230–3304  
***MTTL2*** Transfer RNA, mitochondrial, leucine 2 (CUN) mtDNA 12266–12336  
***MTTM*** Transfer RNA, mitochondrial, methionine mtDNA 4402–4469  
***MTTT*** Transfer RNA, mitochondrial, threonine mtDNA 15888–15953  
***MTTY*** Transfer RNA, mitochondrial, tyrosine mtDNA 5826–5891  
***MyHC*** *myosin Heavy-chain*  
**N O P Q R S T U V**  
***NOS3*** Nitric oxide synthase 3 (endothelial cell) 7q36  
***NPY*** Neuropeptide Y 7p15.1  
***PAI1*** Plasminogen activator inhibitor 1 7q21.3-q22  
***PFKM*** Phosphofructokinase, muscle 12q13.3  
***PGAM2*** Phosphoglycerate mutase 2 (muscle) 7p13-p12  
***PGK1*** Phosphoglycerate kinase 1 Xq13  
***PHKA1*** Phosphorylase kinase, alpha 1 (muscle) Xq12-q13  
***PON1*** Paraoxonase 1 7q21.3  
***PPARA*** Peroxisome proliferative activated receptor, alpha 22q13.31  
***PPARG*** Peroxisome proliferative activated receptor, gamma 3p25  
***PYGM*** Phosphorylase, glycogen, muscle 11q12-q13.2  
***RYR2*** Ryanodine receptor 2 (cardiac) 1q42.1-q43  
***SGCA*** Sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein) 17q21  
***S100A1*** S100 calcium binding protein A1 1q21  
***SUR*** Sulfonylurea receptor 11p15.1  
***TGFB1*** Transforming growth factor beta 1 19q13.2  
***UCP2*** Uncoupling protein 2 11q13  
***UCP3*** Uncoupling protein 3 11q13  
***VDR*** Vitamin D (1,25-dihydroxyvitamin D3) receptor 12q12-q14

The gene symbols, names and cytogenetic locations are from the Locus Link web site available from the National Center for Biotechnology Information (NCBI). For mitochondrial DNA, locations are from the human mitochondrial genome data base.

**TABLE 5. Endurance phenotypes and case-control studies (DNA polymorphisms).**

Gene	Location	Athletes			Controls		
		N	Sports	Freq.	N	Freq.	P
ADRA2A	10q24-q26	140	Endurance	6.7/6.7: 0.77	141	6.7/6.7: 0.62	0.037
				6.7/6.3: 0.21		6.7/6.3: 0.34	
				6.3/6.3: 0.02		6.3/6.3: 0.04	
				6.7: 0.88		6.7: 0.8	
				6.3: 0.12		6.3: 0.2	
ACE	17q23	64	Endurance	II: 0.30	118	II: 0.18	0.03
				ID: 0.55		ID: 0.51	
				DD: 0.16		DD: 0.32	
				I: 0.57		I: 0.43	
				D: 0.43		D: 0.57	
		79	Running	I: 0.57	Ref.	I: 0.49	0.039
				D: 0.43	Pop.	D: 0.51	
		25	Mountain- eering	NA	Ref.	NA	0.02
					Pop.		0.003
		60	Elite athletes (cycling, running, handball)	II: 0.25	Ref.	II: 0.16	0.0009
		56	Elite swimmers (subsample of 103 swimmers)	ID: 0.58	Pop.	ID: 0.45	0.004
				DD: 0.17		DD: 0.39	
				I: 0.54		I: 0.38	
				D: 0.46		D: 0.62	
				II: 0.15		II: 0.24	
				ID: 0.39		ID: 0.49	
				DD: 0.46		DD: 0.27	
				I: 0.34		I: 0.48	
				D: 0.66		D: 0.52	

Reference: Perusse et al. 2003 "The human gene map for performance and health-related fitness phenotypes: the 2002 update" *Med. Sci. Sports Exerc.* 35: 1248-1264.

**TABLE 6.** Genotype and allelic frequencies of *ACTN3* 577R/X alleles in human populations.

Ethnic group	No. of chromosomes	No. of genotypes		Relative allele frequency of 577X
		RX	XX	
Asian	56	14	7	0.5±0.07
Javanese	96	28	12	0.54±0.05
Native American	14	2	2	0.43±0.14
Asia/Americas	166	44	21	0.52±0.04
Hispanic	64	16	5	0.41±0.06
White	214	47	21	0.42±0.03
Europe	278	63	26	0.41±0.03
Aboriginal Australian	174	33	9	0.29±0.03
PNG Highlander	78	16	6	0.36±0.05
Australasia	252	49	15	0.31±0.03
African American	90	12	6	0.27±0.05
African Bantu	156	14	1	0.10±0.05
Africa	246	56	7	0.16±0.05
Unknown	152	50	11	0.47
Total	1094	232	80	